

# CHANGES FROM NORMALITY IN PATIENTS WITH DOWN SYNDROME: AN INTEGRATIVE LITERATURE REVIEW

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**Abstract:** Patients with Down syndrome commonly exhibit deviations from physiological patterns, which may result from genetic, behavioral and environmental factors, leading to dental anomalies, delayed tooth eruption, malocclusions, increased predisposition to dental caries, tooth sensitivity and the presence of fissured tongue. This study aims to gather and synthesize evidence on changes in normality in patients with Down syndrome, providing a comprehensive understanding of the subject. To develop this integrative literature review, the following databases were used: U.S. National Library of Medicine (Pubmed), Virtual Health Library (BVS) and Scientific Electronic Library Online (SciELO), including articles and papers that deal with the presented theme published in the last 10 years, in Portuguese, English and Spanish, that addressed the proposed theme. The studies analyzed demonstrate that individuals with this condition present oral alterations that compromise both masticatory function and aesthetics, thus requiring detailed dental planning, with a focus on functional rehabilitation and improving self-esteem. Thus, understanding these changes becomes essential to ensure adequate management and treatment by health professionals, especially dentists, since hygiene difficulties, dental problems and a greater predisposition to periodontal diseases are present in this population.

**Keywords:** Down syndrome; oral manifestations; malocclusions.

## INTRODUCTION

Down Syndrome (DS) is a genetic condition caused by trisomy 21, being the most common chromosomal alteration in humans. Individuals with DS exhibit a set of particular phenotypic characteristics that include differences in physical, cognitive, and metabolic development. In addition, the syndrome is often associated with various health conditions, such as congenital heart malformations, endocrine disorders, immunodeficiencies, and delayed neuropsychomotor development. (TAVARES, 2022)

One of the most relevant issues in the clinical approach to Down Syndrome is the existence of



changes in what is considered normal for individuals without the condition. Physiological parameters, growth patterns, motor and cognitive development, as well as immune and metabolic responses, can differ significantly compared to the general population. These variations require careful analysis to avoid misdiagnoses and to ensure adequate and individualized medical follow-up. (VILELA, 2018)

The dental surgeon is responsible for each individualized treatment, recognizing the difficulty of each one and correcting irregularities in the oral cavity. Patients with Down syndrome often have changes in oral normalities, which may include changes in the anatomy of the oral cavity, hygiene difficulties, dental problems, and a greater predisposition to periodontal diseases. These changes are consequences of both genetic factors and behavioral and physiological aspects specific to this population. (PELISSON, 2023)

Thus, understanding the changes in normality in patients with DS is essential to optimize health care and improve the quality of life of these individuals. This article aims to review the literature on the main alterations observed in the oral cavity of these patients, such as: tooth absence, changes in enamel and tooth shape, analyzing their dental implications.

## **METHODOLOGICAL PROCEDURES**

This research is an integrative literature review, which aims to gather and synthesize evidence on changes in normality in patients with Down Syndrome, providing a comprehensive understanding of the subject. Searches were carried out in the following databases: U.S. National Library of Medicine (Pubmed), Virtual Health Library (VHL) and Scientific Electronic Library Online (SciELO), using the following descriptors: “Down Syndrome”, “Oral Manifestations” and “Malocclusion” combined with the Boolean operator AND.

For the results, the following inclusion criteria were adopted: articles published in the last 10 years (2015 to May 2025), available in Portuguese, English or Spanish, and works that deal with the theme presented. The exclusion criteria adopted were: incomplete works, published before 2015,



duplicate articles, monographs, and studies that did not meet the eligibility criteria.

## FINDINGS

Title	Authors and year	Goal	Methodology/ Key findings
Oral Alterations in Patients with Down Syndrome: Literature Review	Melo ACFT de. (2022)	To verify information, through a literature review, about patients with Down Syndrome, ranging from oral characteristics that influence dental treatment to genetic alterations.	The dental surgeon must consider two important aspects when treating patients with Down Syndrome: first, the need for a psychologically favorable environment for care; second, the importance of understanding the patient's systemic conditions so as not to compromise homeostasis.
Oral Characteristics and Performance of the Dental Surgeon in the Care of Patients with Down Syndrome.	Vilela JMV, Nascimento MG, Nunes J, Ribeiro. (2018)	To review the literature in order to describe and characterize the conditions that most affect patients with this syndrome.	Patients with this syndrome have a higher prevalence of periodontal disease than normal patients, on the other hand, they have a low prevalence of caries, filled and missing teeth.
Oral Alterations in Patients with Down Syndrome	Pelisson KAM, Borges TS, Villibor FF. (2023)	Demonstrate the main oral alterations found in people with DS.	Down syndrome (DS) is a genetic alteration that influences the individual's life because it affects their development, and can cause systemic and cognitive changes.
Down syndrome. Nature Reviews Disease Primers.	Antonarakis, S. E. et al (2020).	Integrate the current knowledge about Down syndrome (DS), addressing from genetic aspects to clinical manifestations, the use of experimental models and therapeutic perspectives.	The article also highlights recent advances in research aimed at identifying therapeutic interventions capable of improving cognitive function and quality of life in people with DS, with some clinical trials already underway.
Oral health in children with Down syndrome: Parents' views on dental care in Flanders.	Descamps, I., and Marks, L. A. (2015).	To evaluate the opinions and knowledge regarding dental care of parents who have a child with Down syndrome (DS).	explores parents' perceptions of the oral health of their children with Down syndrome and the dental care they receive in the Flanders region, Belgium.



Impact of oral conditions of children/adolescents with Down syndrome on their families' quality of life.	Carrada, C. F., Scalioni, F. A. R., Abreu, L. G., Ribeiro, R. A., Paiva, S. M. (2020).	to evaluate the impact of oral conditions of children and adolescents with Down syndrome (DS) on oral health-related quality of life.	Although the overall impact of oral conditions on the quality of life of families did not differ between groups, certain specific oral conditions in children and adolescents with DS are associated with a worse perception of oral health-related quality of life by their family members.
Oral characteristics and the role of the dental surgeon in the care of patients with Down syndrome.	Vilela JMV, Nascimento MG, Nunes J, Ribeiro EL. (2018)	to review the main oral characteristics of individuals with Down syndrome and discuss the role of the dental surgeon in the care of these patients.	Factors such as plaque, dental calculus, malocclusion, and immune deficiency contribute to the development of periodontal diseases.
Down syndrome: inclusion in municipal dental care.	Nacamura CA, Yamashita JC, Busch RMC, Marta SN. (2015)	OBJECTIVE: To analyze the factors related to the dental care received by children and adolescents with Down syndrome (DS) in the city of Bauru, São Paulo.	The study highlights the need for integrated and multidisciplinary care, involving different dental specialties to promote the effective inclusion of these patients in municipal oral health services.
Oral health problems and their management in patients with Down Syndrome—a narrative review.	Torlińska-Walkowiak, N., Huang, Y. W., Wyzga, S., Opydo-Szymaczek, J. (2024).	consolidate information on the dental challenges faced by individuals with DS, as well as the approaches employed for the care and treatment of these conditions.	The study emphasizes the importance of regular dental evaluations, early interventions such as physical therapy and behavioral management, and a collaborative approach involving oral health professionals, caregivers, and individuals with DS themselves.
Dental care for the patient with Down syndrome.	Pilcher, E. (1998).	to highlight the orofacial and medical particularities associated with DS that directly influence the planning and delivery of dental care.	emphasizes the high incidence of periodontal disease in individuals with DS, often associated with factors such as muscle hypotonia, chronic mouth breathing, immunodeficiency, and ineffective oral hygiene.



## DISCUSSION

Down syndrome is a change in genetic condition caused by the presence of an extra copy of chromosome 21, which results in a total of 47 chromosomes instead of the usual 46. This affects a person's physical and intellectual development. People with Down syndrome may have distinct physical characteristics, such as a shorter neck and greater flexibility in the joints and almond-shaped eyes. In addition, they may have slower cognitive development. (ANTONARAKIS, ET AL. 2020)

Analyzing the oral profile of patients with Down Syndrome, we observed that the most frequent clinical alterations found are: irregular anatomy of the teeth; delayed tooth eruption, often caused by altered bone development; malocclusion, such as anterior open bite (when associated with a deleterious habit) or crossbite; dental agenesis, especially of permanent teeth, which can affect aesthetics and chewing; enamel hypoplasia, where there is a decrease in the amount of enamel, a condition that favors the appearance of caries disease and greater predisposition to tooth sensitivity. (DESCAMPS, MARKS. 2015) These changes may be associated with genetic and environmental factors.

In this way, the role of the dental surgeon during the care of SD patients goes far beyond prevention. It is necessary for your treatment plan to contain the analysis of the facial profile, craniofacial measurements, symmetry and aesthetics as a whole, taking care that the anomalies are corrected so that the patient's self-esteem can be repaired. (CARRADA, ET AL. 2020)

Other common characteristics are a fissured tongue, favoring the accumulation of food in the fissures; macroglossia, change in the size of the tongue that can affect speech, chewing and swallowing; periodontitis associated with poor oral hygiene, which increases the risk of oral infections, and muscle hypotonia, a decrease in muscle tone that can cause weakness. (VILELA, ET AL. 2018; NACAMURA, ET AL. 2015)

In addition, patients with DS may present motor, behavioral, cognitive and other significant systemic characteristics, which challenges the work of the professionals who follow them up, and



multidisciplinary care is essential so that the patient has the opportunity to have a holistic and trained care that corresponds to their specificities. (TORLIŃSKA-WALKOWIAK, ET AL. 2024)

Finally, the oral conditions found in DS patients can substantially affect their quality of life, requiring an attentive, early and individualized look from doctors, dentists and health professionals as a whole, in addition to periodic interventions and preventions, in order to offer care and attention effectively. (PILCHER, 1988)

## **FINAL CONSIDERATIONS**

Down Syndrome presents a series of systemic and oral alterations that require extra attention from health professionals, especially dentists. Changes in the oral cavity, such as dental agenesis, enamel hypoplasia, malocclusions, macroglossia, and predisposition to periodontal disease, reflect not only genetic characteristics, but also the behavioral and physiological challenges of these patients.

Understanding these specificities is essential for planning a humanized, effective treatment adapted to individual needs. The dentist's performance must go beyond the technical aspect, involving an empathetic and multidisciplinary approach that favors the promotion of oral health and quality of life. Therefore, it is essential to continuously invest in professional training and inclusive public policies, ensuring full access to dental care for this population.

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